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ABSTRACT

Reviewed are chromosomal anomalies affecting one's eyes. Brief descriptions are given of the genetic etiology of bilateral retinoblastoma (malignant tumors), aniridia (absence of the iris), cataracts, congenital glaucoma, Reginitis Pigmentosa (progressive deterioration of the visual cells), Choroidermia (degeneration of the vascular coat of the eye), and vitelliform degeneration of Best (degeneration of central vision abilities). The importance of genetic counseling for people with these disorders is stressed. (CL)

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Genetic Factors of Ophthalmic Importance

by

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P50 Session 191

There are numerous conditions which have a genetic basis for their etiology. The eye appears to be one of the organs most commonly affected by hereditary influences. The eye is unique in that it is readily accessable for study with non-invasive techniques. Without performing a biopsy, we can look at the various ocular tissues and diagnose numerous chromosomal anomalies.

Retinoblastoma is the most common malignant ocular tumor of childhood. Its frequency is 1:17,000 to 1:34,000 births. 10% are familial and 90% are sporadic. Of the sporadic cases, 10% are germinal mutations in which the change has occurred in the ova or sperm. The other 90% are somotatic mutations in which the change occurred in the retinal cells of the eye. 30% of cases are bilateral, and 70% are unilateral. It is felt by most authorities in the field that the bilateral cases are due to germinal mutations and are transmitted by an autosomal dominant pattern. Approximately 40% of the offspring of a bilateral retinoblastoma patient will have retinoblastoma as the penetrance is around 80%. There is no doubt that unilateral retinoblastoma can be a germinal mutation as some cases of unilateral retinoblastoma have offspring with unilateral as well as bilateral retinoblastoma. If one child with unilateral retinoblastoma has been born to a family without a history of the disease, there is a 5% chance of a subsequent sibling having retinoblastoma. If a

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child with bilateral retinoblastoma is born to normal parents, there is a 10% chance of a subsequent sibling having retinoblastoma. It is felt that unilateral retinoblastoma has a 15-25% chance of spreading the disease to an offspring. Retinoblastoma has been reported in children with a partial deletion of the long arm of chromosome # 13 or 14. As this is a highly malignant disease, genetic counseling is most important and plays a vital role in the management of these cases. The most common age for presentation is 18 months but several cases have been reported at birth.

Aniridia is a condition in which only a rudimentary iris is present being hidden behind the corneoscleral margin. The influence of heredity is extraordinarilly well marked, the anomaly behaving as a strongly dominant characteristic. A recessive inheritance has occasionally been reported. Sporadic cases appear to occur even more frequently than the hereditary cases. As few as 1:73 or as many as 3:9 cases of sporadic aniridia are associated with Wilms' tumor of the kidney. This malignant tumor of the kidney occurs at an average of 1.8 years in Children with sporadic aniridia. There has also been reported one case of Wilms' tumor in a child with hereditary congenital aniridia. Once a child is born with this defect he must carefully be followed for his first 4 years of life for the possible development of this malignant tumor of the kidney.

The eye has a lens which does the focusing for the visual system just like a camera which also has a focusing lens. Normally the lens is clear but when it becomes opacified it is called a cataract. Most

cataracts occur in adults but occasionally children are born with cataracts. Viral infections such as Rubella can cause cataracts as can syphilis. Neonatal hypocalcemia has been associated with congenital cataracts and the incidence of lenticular opacities in the children of diabetic or pre-diabetic mothers is relatively high. Cataracts also have been associated with chromosomal abnormalities such as in Down's syndrome, Trisomy 13-15, Trisomy 16-18, and Turner's syndrome. It also has been seen in galactosemia caused by either galactose-1-phosphate uridyl transferase deficiency or galactokinase deficiency. Both of these enzyme deficiencies are transmitted as an autosomal recessive disorder.

Congenital glaucoma may be inherited as an autosomal recessive with variable penetrance. Typical signs are present at birth in 35% of patients, at 6 months of age in over 70% and by 12 months of age in approximately 80%. About 75% occur bilaterally. Photophobia, blepharospams, tearing, corneal edema and corneal enlargement are the hallmarks of this disease. Unless the elevated pressure of congenital glaucoma is normalized, vision is lost due to optic nerve atrophy and scarring of the corneas.

Retinitis Pigmentosa is a disease characterized by progressive deterioration of the visual cells, pigment epithelium and choroid. Clinically, a thinning of the retinal vessels, waxy pallor of the optic nerve and the appearance initially at the equator of bone corpuscle like pigment. The condition is always bilateral in familial cases

but sporadic unilateral cases have been noted. The pigmentary change typically becomes visible during the first decade of life and may begin as fine dots which gradually assume the spidery bone-corpuscle appearance. Autosomal recessive is the most common, then autosomal dominant and last is sex-linked recessive which is the most disabling. Usually serious symptoms begin to be apparent in school life (6-12 years), and by 20 years of life they may begin to be incapacitating. The age at which blindness occurs varies. In a large number central vision fails between 40 and 45 years; it frequently lasts until 50 but rarely beyond 60.

Choroidermia is a sex-linked disorder characterized by choroidal degeneration. The choroid is the vascular coat of the eye. Also the pigment epithelium of the retina atrophies. The disease begins in the first decade of life with the chief complaint of night blindness. Good central visual acuity may be maintained for 50 years or more but commonly blindness occurs around age 40.

Vitelliform degeneration of Best is an autosomal dominant disorder that can be congenital or have an onset as late as 7 years of age. This disorder affects the macula which is the central part of the retina and is the part which must function in order to have 20/20 vision. Juvenile macular degeneration of Stargardt occurs between the age of 6 and 20. It usually exhibits autosomal recessive inheritance but autosomal dominant families have been observed. These hereditary macular degenerations while not leading to blindness, do cause serious disability as central vision is lost.

We must give consideration to genetic counseling for families with these various disorders. All of the above diseases inflict considerable harm to the patients who develope each entity. Retinoblastoma can be fatal as well as the Wilms' tumor associated with aniridia. Macular degeneration as well as cataracts can lead to considerable disability even if blindness is not produced. Retinitis Pigmentosa, choroideremia and congenital glaucoma can lead to total blindness. Many of these patients grow up and meet others with hereditary ocular defects at schools for the blind. When they marry, the genes become compounded in their expression in the offspring. The parents as well as the affected offspring should receive genetic counseling concerning their specific disease.